

Claims

1. An isolated DNA sequence characterized by comprising the sequence id. no. 1 or a functional fragment or variant thereof encoding a protein having the same functional activity, or an functionally equivalent isolated DNA sequence hybridizable thereto.

2. An isolated DNA sequence according to claim 1, characterized in that it is associated with diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

3. An isolated DNA-sequence according to claim 1 ~~or 2~~, characterized in that it includes a gene defect responsible for APECED.

4. A DNA sequence according to claim 1, characterized by having the sequence according to sequence id. no 1 or a functional fragment thereof having the sequence according to sequence id. no 3 or sequence id. no 5.

5. A protein characterized by comprising the amino acid sequence id. no. 2 or a functional fragment or variant thereof having the same functional properties.

6. A protein according to claim 5, characterized in that it is associated diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

7. A protein according to claim 5 ~~or 6~~, characterized by having the amino acid sequence id. no. 2, or a functional fragment thereof having the sequence according to sequence id. no. 4, or a functional fragment thereof having the sequence according to sequence id. no 6.

8. A protein according to ~~any of claims 5 to 7~~ ^{claim 5}, characterized by having distinct structural motifs, including the PHD finger motif (PHD), the LXXLL motif (L), proline-rich region (PRR), and cystein-rich region (CRR).

9. A method for the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, characterized by detecting in a biological specimen the presence of a DNA sequence comprising the sequence id. no. 1 or a functional fragment or variant thereof encoding a protein having the same functional activity, or a functionally equivalent isolated DNA-sequence hybridizable thereto.

10. A method according to claim 9, characterized in that the DNA sequence is associated with APECED.

11. A method according to claim 9 ~~or 10~~, characterized in that the DNA sequence includes a gene defect responsible for APECED.

12. A method according to claim 11, characterized in that the gene defect to be detected includes a "C" to "T" transition resulting in the "Arg" to "Stop" nonsense mutation at amino acid position 257 and/or a "A" to "G" transversion resulting in the "Lys" to "Glu" missense mutation at amino acid position 42.

13. A method according to ~~any one of claims 9 to 12~~, characterized in that DNA techniques are used for the detection. *claim 9*

14. A method according to ~~any one of claims 9 to 13~~, characterized in that the detection takes advantage of TaqI or another enzyme cleaving at recognition site 5'-TCGA-3' digestion. *claim 9*

15. A method according to ~~any one of claims 9 to 14~~, characterized in that the disease is autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

16. A method for the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, characterized by detecting in a biological specimen the presence or the absence of a protein comprising the sequence id. no. 2, or a functional fragment thereof having the sequence according to sequence id. no. 4, or a functional fragment thereof having the sequence according to sequence id. no. 6.

17. A method according to claim 16, characterized in that the protein is associated with APECED.

18. A method according to claim 16 ~~or 17~~, characterized in that the disease is autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

19. The use of the DNA sequence according to ~~any one of claims 1 to 4~~ in the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED). *claim 1*

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20. The use of the protein according to ~~any one of claims 5 to 7~~ ^{claim 5} in the diagnosis of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED). ^{claim 1}

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21. The use of the DNA sequence according to ~~any one of claims 1 to 4~~ for the preparation of a medicament useful in a gene therapy method of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED). ^{claim 1}

10 22. The use of the DNA sequence according to ~~any one of claims 1 to 4~~ in the treatment of diseases related to immune maturation and regulation of immune response towards self and nonself, such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED).

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23. Reagents reacting with the DNA sequence according to any one of claims 1 to 4 or the protein of ^a any one of the claims 5 to 8 or with reagents reacting therewith.

24. Reagents according to claim 23, characterized in that they are antibodies.